

# **ADVISORY COMMITTEE ON HERITABLE DISORDERS IN NEWBORNS AND CHILDREN**

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**BETHESDA MARRIOTT HOTEL-POOKS HILL  
BETHESDA, MARYLAND  
SEPTEMBER 24-25, 2009**

## **THURSDAY, SEPTEMBER 24, 2009**

**7:30 A.M. – 8:30 A.M.                      CONTINENTAL BREAKFAST (Committee Members and Presenters)**

**8:30 A.M. – 8:45 A.M.                      WELCOME**

- Approval of Minutes from May 12, 2009 Meeting
- Committee Correspondence

**R. RODNEY HOWELL, M.D.**  
Committee Chair

**8:45 A.M. – 11: 30 A.M.                      SUBCOMMITTEE MEETINGS**

- Follow-up and Treatment
- Education and Training
- Laboratory Standards

**11:30 A.M. –11:45 A.M.                      BREAK**

**11:45 A.M. – 12:30 P.M.                      SUBCOMMITTEE REPORTS AND DISCUSSION**

- Subcommittee on Laboratory Standards and Procedures

**Gerard Vockley, M.D., PH.D.**  
Committee Member

- Subcommittee on Education and Training

**Jana Monaco and Tracy L. Trotter, M.D.**  
Committee Member

- Subcommittee on Follow-up and Treatment

**Coleen Boyle, PH.D., M.S.**  
Committee Member

**12:30 P.M. – 1:15 P.M.                      LUNCH (Committee Members and Presenters)**

<b>1:15 P.M. – 2:00 P.M.</b>	<b>THE NATIONWIDE HEALTH INFORMATION NETWORK</b>  <b>Charles Friedman, M.D.</b> Deputy Director for the Office of the National Coordinator for Health Information Technology
<b>2:00 P.M. – 2:15 P.M.</b>	<b>STATUS OF THE IMPLEMENTATION OF THE NEWBORN SCREENING USE CASE</b>  <b>Alan Zuckerman, M.D.</b> Georgetown University
<b>2:15 P.M. – 2:45 P.M.</b>	<b>NEWBORN SCREENING CODES &amp; TERMINOLOGY AND AN APPROACH TO A STANDARD REPORT PAYLOAD</b>  <b>Kin Wah Fung, M.D.</b> National Library of Medicine
<b>2:45 P.M. – 3:00 P.M.</b>	<b>NEWBORN SCREENING WEB PORTAL CONCEPT</b>  <b>Gregory Downing D.O., Ph.D. and Constanze Coon, Ph.D.</b> Personalized Health Care Initiative, OS/HHS
<b>3:00 P.M. – 3:15 P.M.</b>	<b>BREAK</b>
<b>3:15 P.M. – 4:00 P.M.</b>	<b>MEASURES FOR QUALITY</b>  <b>Sarah Scholle, DR.PH.</b> National Center for Quality Assurance
<b>4:00 P.M. – 4:30 P.M.</b>	<b>NOMINATION OF ALPHA THALASSEMIA: HEMOGLOBIN H DISEASE TO THE ACHDNC’S RECOMMENDED UNIFORM SCREENING PANEL</b>  <b>Kwaku Ohene-Frempong, M.D.</b> Committee Member
<b>4:30 P.M. – 5:00 P.M.</b>	<b>COMMITTEE DISCUSSION</b>
<b>5:00 P.M.</b>	<b>ADJOURN</b>

**FRIDAY, SEPTEMBER 25, 2009**

<b>7:30 A.M. – 8:30 A.M.</b>	<b>CONTINENTAL BREAKFAST (COMMITTEE MEMBERS AND PRESENTERS)</b>
<b>8:30 A.M. – 9:00 A.M.</b>	<b>USE AND STORAGE OF RESIDUAL BLOOD SPOTS WORKGROUP: BRIEFING PAPER REPORT</b>  <b>Jana Monaco</b> Committee Member  <b>Brad Therrell, PH.D.</b> <b>Harry Hannon, PH.D.</b> National Newborn Screening and Genetic Resource Center
<b>9:00 A.M. – 9:30 A.M.</b>	<b>COMMITTEE DISCUSSION: BRIEFING PAPER</b>  <b>R. Rodney Howell, M.D.</b> Committee Chair
<b>9:30 A.M. – 10:00 A.M.</b>	<b>PUBLIC COMMENTS</b>
<b>10 A.M. – 10:30 A.M.</b>	<b>BREAK</b>
<b>10:30 A.M. – 11:30 A.M.</b>	<b>EVIDENCE REVIEW WORKGROUP: FINAL DRAFT REPORT ON THE CANDIDATE NOMINATION OF KRABBE DISEASE</b>  <b>James M. Perrin, M.D.</b> Evidence Review Workgroup Chairperson
<b>11:30 A.M. – 12:00 P.M.</b>	<b>PUBLIC COMMENTS</b>
<b>12:00 P.M. – 1:00 P.M.</b>	<b>LUNCH (Committee Members and Presenters)</b>
<b>1:00 P.M. – 2:00 P.M.</b>	<b>COMMITTEE DISCUSSION ON THE CANDIDATE NOMINATION OF KRABBE DISEASE</b>  <b>Piero Rinaldo, M.D., PH.D.</b> Committee Member
<b>2:00 P.M. – 2:30 P.M.</b>	<b>COMMITTEE REPORT FOR CONGRESS</b> <b>POLICY PAPER: NEWBORN SCREENING AND HEALTH CARE REFORM</b>  <b>R. Rodney Howell, M.D.</b> Committee Chair
<b>2:30 P.M. – 3:00 P.M.</b>	<b>COMMITTEE BUSINESS</b> <ul style="list-style-type: none"><li>• Calendar for 2010 Committee Meetings</li><li>• Agenda Items For January 2010 Meeting</li></ul> <b>R. Rodney Howell, M.D.</b> Committee Chair
<b>3:00 P.M.</b>	<b>ADJOURN</b>